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Cognitive function in Rett syndrome: profoundly impaired or near normal?

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Rett syndrome (RTT) is a rare neurodevelopmental disorder that predominantly affects girls. Following a short period of apparently near-normal development, the children regress, with most losing any acquired speech and functional hand-use, the latter being replaced by characteristic hand stereotypies. The traditional view has held that children with RTT are profoundly cognitively impaired; typical estimates have suggested a mental development of around 8-12 months. Despite this, parents often insist that their child shows a good level of understanding, highlighting the very intense eye contact and apparent communication by eye-pointing seen in many of the children (e.g. Urbanowicz et al., 2016). Cognitive assessment is undoubtedly challenging in individuals with profound physical disabilities, especially in those without purposeful hand-use or speech. As a consequence, conventional testing methods, which require motor or verbal responses or both, are inappropriate for children with RTT. Recent advances in eye-gaze (or eye-tracking) technology, however, offer a way for people with RTT to make choices and, moreover, communicate independently. Djukic and colleagues (2012) made the intriguing observation that children with RTT explore novel stimuli with their eyes in a similar way to neurotypical children, suggesting that they derived meaning from the stimuli in a comparable way to their peers. While this seemed to support the perception of

parents and questioned a low mental development, it was based on a somewhat indirect and passive measurement.

In a new paper in this issue of the European Journal of Paediatric Neurology, Ahonniska-Assa and colleagues aimed to address cognitive abilities of girls with RTT more directly. They modified a standard test of receptive vocabulary, the Peabody Picture Vocabulary Test, in a group of 17 girls with RTT. While this test does not require a verbal response, participants are required to indicate, by pointing, which of four pictures is associated with a prompt given by the tester. Ahonniska et al. displayed the four pictures instead on a computer monitor. Subjects then indicated their response by looking at one of the pictures. This response was registered by the eye gaze system. The study found that almost a third of the girls with RTT tested showed receptive vocabulary that was at a level indicative of either mild cognitive impairment (2/17) or even within the normal range (4/17). This is a remarkable finding that has strong implications for our understanding of the cognitive abilities in Rett syndrome.

Could this test have over-estimated the abilities in RTT? Given the testing protocol, this seems unlikely. However, there remains the possibility that the findings underestimate the potential of girls with RTT. Eye gaze is not an easy technique to master. Most of the girls were not routinely using eye gaze devices at the start of the study and the normative data are for children who did not have to use eye gaze. Moreover, the exposure to language, and certainly the educational input, may very well differ in children where there has been a presumption of severe cognitive impairment. Taken together, these factors may account for the range of abilities

measured in the children with RTT; the low scores for some of the children may represent an underestimation of their abilities.

The results of this study have profound implications for the future of girls and women with RTT, who were previously thought to be at the very lowest end of the cognitive spectrum. If patients with RTT are functioning at a level of mild cognitive impairment, or even within the normal range of intelligence, educational strategies, communication support and daily routines need to be revisited. Eye-tracking and gaze-controlled communication may be the key to unlocking the potential of people with Rett syndrome.

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